S Rahman

List of Publications by Year
in descending order

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174 papers	12,066 citations	26630 56 h-index	30087 103 g-index
185 all docs	185 docs citations	185 times ranked	13091 citing authors

#	Article	IF	Citations
1	Quo vadis now: Beyond genomics to an era of personalised medicine. Journal of Inherited Metabolic Disease, 2022, 45, 129-131.	3.6	0
2	Research priorities for mitochondrial disorders: Current landscape and patient and professional views. Journal of Inherited Metabolic Disease, 2022, 45, 796-803.	3.6	5
3	Moving towards clinical trials for mitochondrial diseases. Journal of Inherited Metabolic Disease, 2021, 44, 22-41.	3.6	45
4	Comment on â€œA severe linezolidâ€induced rhabdomyolysis and lactic acidosis in Leigh syndromeâ€• Journal of Inherited Metabolic Disease, 2021, 44, 6-7.	3.6	2
5	Seeking impact: Global perspectives on outcome measure selection for translational and clinical research for primary mitochondrial disorders. Journal of Inherited Metabolic Disease, 2021, 44, 343-357.	3.6	13
6	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	3.6	146
7	Editorial: Mitochondrial medicine special issue. Journal of Inherited Metabolic Disease, 2021, 44, 289-291.	3.6	3
8	Biparental inheritance of mitochondrial DNA revisited. Nature Reviews Genetics, 2021, 22, 477-478.	16.3	12
9	Diagnosing Mitochondrial Disorders Remains Challenging in the Omics Era. Neurology: Genetics, 2021, 7, e597.	1.9	13
10	Biallelic P4HTM variants associated with HIDEA syndrome and mitochondrial respiratory chain complex I deficiency. European Journal of Human Genetics, 2021, 29, 1536-1541.	2.8	7
11	Effect of neuropsychiatric medications on mitochondrial function: For better or for worse. Neuroscience and Biobehavioral Reviews, 2021, 127, 555-571.	6.1	15
12	Expanding the phenotypic spectrum of BCS1Lâ€related mitochondrial disease. Annals of Clinical and Translational Neurology, 2021, 8, 2155-2165.	3.7	11
13	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	6.0	42
14	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
15	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	2.4	14
16	Simplifying the clinical classification of polymerase gamma (POLG) disease based on age of onset; studies using a cohort of 155 cases. Journal of Inherited Metabolic Disease, 2020, 43, 726-736.	3.6	33
17	The impact of gender, puberty, and pregnancy in patients with POLG disease. Annals of Clinical and Translational Neurology, 2020, 7, 2019-2025.	3.7	7
18	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	3.3	17

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19	Cardiac valve involvement in ADAR-related type I interferonopathy. Journal of Medical Genetics, 2020, 57, 475-478.	3.2	19
20	Mitochondrial disease in children. Journal of Internal Medicine, 2020, 287, 609-633.	6.0	83
21	Nuclear-mitochondrial DNA segments resemble paternally inherited mitochondrial DNA in humans. Nature Communications, 2020, 11, 1740.	12.8	75
22	Safety of drug use in patients with a primary mitochondrial disease: An international Delphiâ€based consensus. Journal of Inherited Metabolic Disease, 2020, 43, 800-818.	3.6	42
23	Bi-allelic Variants in TKFC Encoding Triokinase/FMN Cyclase Are Associated with Cataracts and Multisystem Disease. American Journal of Human Genetics, 2020, 106, 256-263.	6.2	16
24	Erythrocyte Encapsulated Thymidine Phosphorylase for the Treatment of Patients with Mitochondrial Neurogastrointestinal Encephalomyopathy: Study Protocol for a Multi-Centre, Multiple Dose, Open Label Trial. Journal of Clinical Medicine, 2019, 8, 1096.	2.4	39
25	Differential phenotypic expression of a novel PDHA1 mutation in a female monozygotic twin pair. Human Genetics, 2019, 138, 1313-1322.	3.8	12
26	Advances in the treatment of mitochondrial epilepsies. Epilepsy and Behavior, 2019, 101, 106546.	1.7	17
27	Diagnosis of â€˜possibleâ€™ mitochondrial disease: an existential crisis. Journal of Medical Genetics, 2019, 56, 123-130.	3.2	42
28	Disorders of riboflavin metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 608-619.	3.6	82
29	The utility of phenomics in diagnosis of inherited metabolic disorders. Clinical Medicine, 2019, 19, 30-36.	1.9	15
30	B Vitamins: Small molecules, big effects. Journal of Inherited Metabolic Disease, 2019, 42, 579-580.	3.6	6
31	Mutations inELAC2associated with hypertrophic cardiomyopathy impair mitochondrial tRNA 3â€²â€end processing. Human Mutation, 2019, 40, 1731-1748.	2.5	31
32	Cerebral folate deficiency: Analytical tests and differential diagnosis. Journal of Inherited Metabolic Disease, 2019, 42, 655-672.	3.6	69
33	POLG-related disorders and their neurological manifestations. Nature Reviews Neurology, 2019, 15, 40-52.	10.1	229
34	Systems Biology Approaches Toward Understanding Primary Mitochondrial Diseases. Frontiers in Genetics, 2019, 10, 19.	2.3	12
35	The CAPOS mutation in ATP1A3 alters Na/K-ATPase function and results in auditory neuropathy which has implications for management. Human Genetics, 2018, 137, 111-127.	3.8	24
36	Retrospective natural history of thymidine kinase 2 deficiency. Journal of Medical Genetics, 2018, 55, 515-521.	3.2	73

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37	Spectrum of movement disorders and neurotransmitter abnormalities in paediatric POLG disease. Journal of Inherited Metabolic Disease, 2018, 41, 1275-1283.	3.6	12
38	Mitochondrial diseases and status epilepticus. Epilepsia, 2018, 59, 70-77.	5.1	30
39	Near infrared spectroscopy with a vascular occlusion test as a biomarker in children with mitochondrial and other neuro-genetic disorders. PLoS ONE, 2018, 13, e0199756.	2.5	3
40	Outcome measures for children with mitochondrial disease: consensus recommendations for future studies from a Delphiâ€based international workshop. Journal of Inherited Metabolic Disease, 2018, 41, 1267-1273.	3.6	24
41	Natural history of mitochondrial disorders: a systematic review. Essays in Biochemistry, 2018, 62, 423-442.	4.7	30
42	Rapid Paediatric Sequencing (RaPS): comprehensive real-life workflow for rapid diagnosis of critically ill children. Journal of Medical Genetics, 2018, 55, 721-728.	3.2	98
43	Elevated cerebrospinal fluid protein in <scp>POLG</scp>â€related epilepsy: Diagnostic and prognostic implications. Epilepsia, 2018, 59, 1595-1602.	5.1	6
44	Mitochondrial medicine in the omics era. Lancet, The, 2018, 391, 2560-2574.	13.7	197
45	The clinical spectrum and natural history of early-onset diseases due to DNA polymerase gamma mutations. Genetics in Medicine, 2017, 19, 1217-1225.	2.4	45
46	Common data elements for clinical research in mitochondrial disease: a National Institute for Neurological Disorders and Stroke project. Journal of Inherited Metabolic Disease, 2017, 40, 403-414.	3.6	15
47	Leigh map: A novel computational diagnostic resource for mitochondrial disease. Annals of Neurology, 2017, 81, 9-16.	5.3	68
48	The presence of anaemia negatively influences survival in patients with POLG disease. Journal of Inherited Metabolic Disease, 2017, 40, 861-866.	3.6	8
49	International Workshop:. Neuromuscular Disorders, 2017, 27, 1126-1137.	0.6	58
50	Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society. Genetics in Medicine, 2017, 19, 1380-1397.	2.4	173
51	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
52	Recognition, investigation and management of mitochondrial disease. Archives of Disease in Childhood, 2017, 102, 1082-1090.	1.9	32
53	Mutations in SLC25A22: hyperprolinaemia, vacuolated fibroblasts and presentation with developmental delay. Journal of Inherited Metabolic Disease, 2017, 40, 385-394.	3.6	16
54	Mitochondrial disease and endocrine dysfunction. Nature Reviews Endocrinology, 2017, 13, 92-104.	9.6	146

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55	Human COQ9 Rescues a coq9 Yeast Mutant by Enhancing Coenzyme Q Biosynthesis from 4-Hydroxybenzoic Acid and Stabilizing the CoQ-Synthome. Frontiers in Physiology, 2017, 8, 463.	2.8	13
56	A Clinical, Neuropathological and Genetic Study of Homozygous A467T POLG-Related Mitochondrial Disease. PLoS ONE, 2016, 11, e0145500.	2.5	36
57	Leigh syndrome: One disorder, more than 75 monogenic causes. Annals of Neurology, 2016, 79, 190-203.	5.3	374
58	Incidence of Primary Mitochondrial Disease in Children Younger Than 2 Years Presenting With Acute Liver Failure. Journal of Pediatric Gastroenterology and Nutrition, 2016, 63, 592-597.	1.8	40
59	TRNT1 deficiency: clinical, biochemical and molecular genetic features. Orphanet Journal of Rare Diseases, 2016, 11, 90.	2.7	64
60	Advantages and pitfalls of an extended gene panel for investigating complex neurometabolic phenotypes. Brain, 2016, 139, 2844-2854.	7.6	35
61	Neurophysiological profile of peripheral neuropathy associated with childhood mitochondrial disease. Mitochondrion, 2016, 30, 162-167.	3.4	13
62	The pleiotropic effects of decanoic acid treatment on mitochondrial function in fibroblasts from patients with complex I deficient Leigh syndrome. Journal of Inherited Metabolic Disease, 2016, 39, 415-426.	3.6	44
63	Position statement on the role of healthcare professionals, patient organizations and industry in European Reference Networks. Orphanet Journal of Rare Diseases, 2016, 11, 7.	2.7	14
64	Recommendations and guidelines in the JIMD: suggested procedures and avoidance of conflicts of interest. Journal of Inherited Metabolic Disease, 2016, 39, 327-329.	3.6	1
65	Peer review fraudâ€"itâ€™s not big and itâ€™s not clever. Journal of Inherited Metabolic Disease, 2016, 39, 1-2.	3.6	12
66	Extra-ocular muscle MRI in genetically-defined mitochondrial disease. European Radiology, 2016, 26, 130-137.	4.5	24
67	Quo vadis: the reâ€definition of â€œinborn metabolic diseasesâ€• Journal of Inherited Metabolic Disease, 2015, 38, 1003-1006.	3.6	48
68	Biâ€allelic CLPB mutations cause cataract, renal cysts, nephrocalcinosis and 3â€methylglutaconic aciduria, a novel disorder of mitochondrial protein disaggregation. Journal of Inherited Metabolic Disease, 2015, 38, 211-219.	3.6	46
69	TMEM70 deficiency: longâ€term outcome of 48 patients. Journal of Inherited Metabolic Disease, 2015, 38, 417-426.	3.6	51
70	Mitochondrial m.1584A 12S m62A rRNA methylation in families with m.1555A>G associated hearing loss. Human Molecular Genetics, 2015, 24, 1036-1044.	2.9	43
71	Pathophysiology of mitochondrial disease causing epilepsy and status epilepticus. Epilepsy and Behavior, 2015, 49, 71-75.	1.7	56
72	Paediatric single mitochondrial DNA deletion disorders: an overlapping spectrum of disease. Journal of Inherited Metabolic Disease, 2015, 38, 445-457.	3.6	95

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73	Emerging aspects of treatment in mitochondrial disorders. Journal of Inherited Metabolic Disease, 2015, 38, 641-653.	3.6	32
74	Oxidative phosphorylation gene expression falls at onset and throughout the development of meningococcal sepsis-induced multi-organ failure in children. Intensive Care Medicine, 2015, 41, 1489-1490.	8.2	5
75	Tubular aggregates caused by serine active site containing 1 (<scp>SERAC1</scp>) mutations in a patient with a mitochondrial encephalopathy. Neuropathology and Applied Neurobiology, 2015, 41, 399-402.	3.2	10
76	Signal transducer and activator of transcription 2 deficiency is a novel disorder of mitochondrial fission. Brain, 2015, 138, 2834-2846.	7.6	78
77	Can folic acid have a role in mitochondrial disorders?. Drug Discovery Today, 2015, 20, 1349-1354.	6.4	38
78	The urinary proteome and metabonome differ from normal in adults with mitochondrial disease. Kidney International, 2015, 87, 610-622.	5.2	41
79	Recurrent rhabdomyolysis due to muscle Î²-enolase deficiency: very rare or underestimated?. Journal of Neurology, 2014, 261, 2424-2428.	3.6	22
80	Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. European Journal of Human Genetics, 2014, 22, 184-191.	2.8	52
81	Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. Brain, 2014, 137, 44-56.	7.6	143
82	The ketogenic diet component decanoic acid increases mitochondrial citrate synthase and complex I activity in neuronal cells. Journal of Neurochemistry, 2014, 129, 426-433.	3.9	153
83	Development of pharmacological strategies for mitochondrial disorders. British Journal of Pharmacology, 2014, 171, 1798-1817.	5.4	64
84	Gentamicin, genetic variation and deafness in preterm children. BMC Pediatrics, 2014, 14, 66.	1.7	10
85	Brown-Vialetto-van Laere syndrome: A riboflavin responsive neuronopathy of infancy with singular features. European Journal of Paediatric Neurology, 2014, 18, 231-234.	1.6	18
86	Effect of Coenzyme Q10 supplementation on mitochondrial electron transport chain activity and mitochondrial oxidative stress in Coenzyme Q10 deficient human neuronal cells. International Journal of Biochemistry and Cell Biology, 2014, 50, 60-63.	2.8	57
87	G.P.191. Neuromuscular Disorders, 2014, 24, 867.	0.6	0
88	Successful reversal of propionic acidaemia associated cardiomyopathy: Evidence for low myocardial coenzyme Q10 status and secondary mitochondrial dysfunction as an underlying pathophysiological mechanism. Mitochondrion, 2014, 17, 150-156.	3.4	36
89	SURF1 deficiency: a multi-centre natural history study. Orphanet Journal of Rare Diseases, 2013, 8, 96.	2.7	107
90	Coenzyme Q₁₀ quantification in muscle, fibroblasts and cerebrospinal fluid by liquid chromatography/tandem mass spectrometry using a novel deuterated internal standard. Rapid Communications in Mass Spectrometry, 2013, 27, 924-930.	1.5	18

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91	NDUFA4 Mutations Underlie Dysfunction of a Cytochrome c Oxidase Subunit Linked to Human Neurological Disease. Cell Reports, 2013, 3, 1795-1805.	6.4	104
92	Novel Mutations inSCO1as a Cause of Fatal Infantile Encephalopathy and Lactic Acidosis. Human Mutation, 2013, 34, 1366-1370.	2.5	36
93	HIBCH mutations can cause Leigh-like disease with combined deficiency of multiple mitochondrial respiratory chain enzymes and pyruvate dehydrogenase. Orphanet Journal of Rare Diseases, 2013, 8, 188.	2.7	70
94	A distinct mitochondrial myopathy, lactic acidosis and sideroblastic anemia (MLASA) phenotype associates with YARS2 mutations. American Journal of Medical Genetics, Part A, 2013, 161, 2334-2338.	1.2	42
95	PGC-1Î² mediates adaptive chemoresistance associated with mitochondrial DNA mutations. Oncogene, 2013, 32, 2592-2600.	5.9	35
96	The UK MRC Mitochondrial Disease Patient Cohort Study: clinical phenotypes associated with the m.3243A>G mutation–implications for diagnosis and management. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 936-938.	1.9	193
97	Inborn errors of metabolism causing epilepsy. Developmental Medicine and Child Neurology, 2013, 55, 23-36.	2.1	69
98	Mitochondrial diseaseâ€"an important cause of end-stage renal failure. Pediatric Nephrology, 2013, 28, 357-361.	1.7	37
99	Human neuronal coenzyme Q₁₀ deficiency results in global loss of mitochondrial respiratory chain activity, increased mitochondrial oxidative stress and reversal of ATP synthase activity: implications for pathogenesis and treatment. Journal of Inherited Metabolic Disease, 2013, 36, 63-73.	3.6	49
100	Levels of 5-methyltetrahydrofolate and ascorbic acid in cerebrospinal fluid are correlated: Implications for the accelerated degradation of folate by reactive oxygen species. Neurochemistry International, 2013, 63, 750-755.	3.8	17
101	Gastrointestinal and hepatic manifestations of mitochondrial disorders. Journal of Inherited Metabolic Disease, 2013, 36, 659-673.	3.6	53
102	COX10Mutations Resulting in Complex Multisystem Mitochondrial Disease That Remains Stable Into Adulthood. JAMA Neurology, 2013, 70, 1556-61.	9.0	27
103	Distal myopathy with cachexia: an unrecognised phenotype caused by dominantly-inherited mitochondrial polymerase Î³ mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2013, 84, 107-110.	1.9	16
104	Treatable Leigh-like encephalopathy presenting in adolescence. BMJ Case Reports, 2013, 2013, bcr2013200838-bcr2013200838.	0.5	41
105	Genetic dysfunction of MT-ATP6 causes axonal Charcot-Marie-Tooth disease. Neurology, 2012, 79, 1145-1154.	1.1	97
106	Complex I deficiency: clinical features, biochemistry and molecular genetics. Journal of Medical Genetics, 2012, 49, 578-590.	3.2	264
107	Hearing in 44â€"45 year olds with m.1555A>G, a genetic mutation predisposing to aminoglycoside-induced deafness: a population based cohort study. BMJ Open, 2012, 2, e000411.	1.9	40
108	POLG mutations and age at menopause. Human Reproduction, 2012, 27, 2243-2244.	0.9	12

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109	Study of LPIN1, LPIN2 and LPIN3 in rhabdomyolysis and exerciseâ€induced myalgia. Journal of Inherited Metabolic Disease, 2012, 35, 1119-1128.	3.6	75
110	Plasma thiol status is altered in children with mitochondrial diseases. Scandinavian Journal of Clinical and Laboratory Investigation, 2012, 72, 152-157.	1.2	17
111	Single deletions in mitochondrial DNA â€“ Molecular mechanisms and disease phenotypes in clinical practice. Neuromuscular Disorders, 2012, 22, 577-586.	0.6	62
112	Mitochondrial disease and epilepsy. Developmental Medicine and Child Neurology, 2012, 54, 397-406.	2.1	140
113	Further delineation of pontocerebellar hypoplasia type 6 due to mutations in the gene encoding mitochondrial arginylâ€tRNA synthetase, RARS2. Journal of Inherited Metabolic Disease, 2012, 35, 459-467.	3.6	51
114	Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. Journal of Medical Genetics, 2011, 48, 610-617.	3.2	49
115	P47 Mutations in the novel chaperone FOXRED1 cause mitochondrial complex I deficiency. Neuromuscular Disorders, 2011, 21, S19.	0.6	0
116	P53 Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. Neuromuscular Disorders, 2011, 21, S21.	0.6	10
117	P58 Mitochondrial respiratory chain enzyme deficiency expressed during muscle development. Neuromuscular Disorders, 2011, 21, S23.	0.6	0
118	P80 The MRC Centre for Translational Research in Neuromuscular Disease: Mitochondrial Disease Patient Cohort Study UK. Neuromuscular Disorders, 2011, 21, S29-S30.	0.6	2
119	On the Nature of Fear of Falling in Parkinsonâ€™s Disease. Behavioural Neurology, 2011, 24, 219-228.	2.1	84
120	Mutations in the mitochondrial complex I assembly factor NDUFAF1 cause fatal infantile hypertrophic cardiomyopathy. Journal of Medical Genetics, 2011, 48, 691-697.	3.2	64
121	14 Mitochondrial cardiomyopathy caused by defective assembly of respiratory chain complex I. Heart, 2011, 97, e8-e8.	2.9	0
122	On the nature of fear of falling in Parkinson's disease. Behavioural Neurology, 2011, 24, 219-28.	2.1	45
123	Treatment of CoQ10 Deficient Fibroblasts with Ubiquinone, CoQ Analogs, and Vitamin C: Time- and Compound-Dependent Effects. PLoS ONE, 2010, 5, e11897.	2.5	92
124	PONM13 Fatty acid oxidation disorders in adults: a potentially treatable cause of muscle disease. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, e63-e63.	1.9	0
125	PORT03 MRC mitochondrial cohort study: development of a UK database. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, e66-e66.	1.9	0
126	Reactive oxygen species, oxidative stress, and cell death correlate with level of CoQ ₁₀ deficiency. FASEB Journal, 2010, 24, 3733-3743.	0.5	142

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127	The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. Neurology, 2010, 74, 1619-1626.	1.1	84
128	PONM21 Electron microscopy does not add to the diagnostic accuracy of muscle biopsy for suspected mitochondrial disease. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, e65-e65.	1.9	1
129	FOXRED1, encoding an FAD-dependent oxidoreductase complex-I-specific molecular chaperone, is mutated in infantile-onset mitochondrial encephalopathy. Human Molecular Genetics, 2010, 19, 4837-4847.	2.9	79
130	Aminoglycoside-induced deafness during treatment of acute leukaemia. Archives of Disease in Childhood, 2010, 95, 153-155.	1.9	8
131	Corrigendum to â€˜Prevalence and natural history of heart disease in adults with primary mitochondrial respiratory chain diseaseâ€™ [Eur J Heart Fail2010;12:114-121]. European Journal of Heart Failure, 2010, 12, 1017-1017.	7.1	0
132	POG04 Multiple mitochondrial DNA deletions, cyclooxygenase-negative fibres and slowly progressive cognitive decline with psychiatric features. Journal of Neurology, Neurosurgery and Psychiatry, 2010, 81, e49-e49.	1.9	0
133	Cardiac manifestations of mitochondrial disorders: reply. European Journal of Heart Failure, 2010, 12, 637-638.	7.1	0
134	Prevalence and natural history of heart disease in adults with primary mitochondrial respiratory chain disease. European Journal of Heart Failure, 2010, 12, 114-121.	7.1	117
135	P69 MRC mitochondrial cohort study: development of a UK database. Neuromuscular Disorders, 2010, 20, S23.	0.6	0
136	P70 Non-invasive diagnosis of single deletion disorders in children with suspected mitochondrial disease. Neuromuscular Disorders, 2010, 20, S24.	0.6	0
137	P77 Complex I-deficient Leigh syndrome caused by a novel homozygous deletion in NDUFS4. Neuromuscular Disorders, 2010, 20, S25-S26.	0.6	1
138	Diagnosis and therapy in neuromuscular disorders: diagnosis and new treatments in mitochondrial diseases. Journal of Neurology, Neurosurgery and Psychiatry, 2009, 80, 943-953.	1.9	131
139	Analysis of mutant DNA polymerase Î³ in patients with mitochondrial DNA depletion. Human Mutation, 2009, 30, 248-254.	2.5	52
140	Status epilepticus in children with Alpersâ€™ disease caused by POLG1 mutations: EEG and MRI features. Epilepsia, 2009, 50, 1596-1607.	5.1	141
141	A Nonsense Mutation in COQ9 Causes Autosomal-Recessive Neonatal-Onset Primary Coenzyme Q10 Deficiency: A Potentially Treatable Form of Mitochondrial Disease. American Journal of Human Genetics, 2009, 84, 558-566.	6.2	206
142	G.P.3.04 Autosomal dominant Progressive External Ophthalmoplegia (adPEO) due to mutations in the PEO1 gene: A clinical, histochemical and molecular survey of 33 patients. Neuromuscular Disorders, 2009, 19, 562.	0.6	0
143	Prevalence of Mitochondrial 1555Aâ†’G Mutation in European Children. New England Journal of Medicine, 2009, 360, 640-642.	27.0	143
144	Diagnosis of mitochondrial DNA depletion syndromes. Archives of Disease in Childhood, 2009, 94, 3-5.	1.9	60

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145	G.P.3.14 Comparative human mitochondrial genome analysis using the affymetrix Mitochip v2 and conventional cycle sequencing. Neuromuscular Disorders, 2008, 18, 755-756.	0.6	0
146	The Factors that Induce or Overcome Freezing of Gait in Parkinsonâ€™s Disease. Behavioural Neurology, 2008, 19, 127-136.	2.1	140
147	Ototoxicity caused by aminoglycosides. BMJ: British Medical Journal, 2007, 335, 784-785.	2.3	64
148	ALLOGENEIC STEM CELL TRANSPLANTATION CORRECTS BIOCHEMICAL DERANGEMENTS IN MNGIE. Neurology, 2007, 68, 1872-1873.	1.1	6
149	Phenotypic variability of mitochondrial disease caused by a nuclear mutation in complex II. Molecular Genetics and Metabolism, 2006, 89, 214-221.	1.1	42
150	Dominant inheritance of premature ovarian failure associated with mutant mitochondrial DNA polymerase gamma. Human Reproduction, 2006, 21, 2467-2473.	0.9	153
151	Novel TRPM6 Mutations in 21 Families with Primary Hypomagnesemia and Secondary Hypocalcemia. Journal of the American Society of Nephrology: JASN, 2005, 16, 3061-3069.	6.1	157
152	Deficiency of the ADP-Forming Succinyl-CoA Synthase Activity Is Associated with Encephalomyopathy and Mitochondrial DNA Depletion. American Journal of Human Genetics, 2005, 76, 1081-1086.	6.2	284
153	New phenotypic diversity associated with the mitochondrial tRNASer(UCN) gene mutation. Neuromuscular Disorders, 2005, 15, 364-371.	0.6	29
154	Ethylmalonic Encephalopathy Is Caused by Mutations in ETHE1, a Gene Encoding a Mitochondrial Matrix Protein. American Journal of Human Genetics, 2004, 74, 239-252.	6.2	192
155	Mutation and biochemical analysis in carnitine palmitoyltransferase type II (CPT II) deficiency. Journal of Inherited Metabolic Disease, 2003, 26, 543-557.	3.6	47
156	Mitochondrial HMG-CoA synthase deficiency: identification of two further patients carrying two novel mutations. European Journal of Pediatrics, 2003, 162, 279-280.	2.7	28
157	Genetic and functional analyses of FH mutations in multiple cutaneous and uterine leiomyomatosis, hereditary leiomyomatosis and renal cancer, and fumarate hydratase deficiency. Human Molecular Genetics, 2003, 12, 1241-1252.	2.9	272
158	Alpers Syndrome with Mitochondrial Dna Depletion. Clinical Science, 2002, 103, 51P-51P.	0.0	0
159	Alpers syndrome with mitochondrial DNA depletion. Neuropathology and Applied Neurobiology, 2002, 28, 160-160.	3.2	0
160	Germline mutations in FH predispose to dominantly inherited uterine fibroids, skin leiomyomata and papillary renal cell cancer. Nature Genetics, 2002, 30, 406-410.	21.4	1,426
161	Diagnostic Value of Succinate Ubiquinone Reductase Activity in the Identification of Patients with Mitochondrial DNA Depletion. Journal of Inherited Metabolic Disease, 2002, 25, 7-16.	3.6	26
162	Neonatal presentation of coenzyme Q10 deficiency. Journal of Pediatrics, 2001, 139, 456-458.	1.8	112

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163	Decrease of 3243 Aâ†'G mtDNA Mutation from Blood in MELAS Syndrome: A Longitudinal Study. American Journal of Human Genetics, 2001, 68, 238-240.	6.2	226
164	A SURF1 gene mutation presenting as isolated leukodystrophy. Annals of Neurology, 2001, 49, 797-800.	5.3	78
165	Early onset of complete heart block in Pearson syndrome. Journal of Inherited Metabolic Disease, 2000, 23, 753-754.	3.6	7
166	Cytochrome oxidase immunohistochemistry: clues for genetic mechanisms. Brain, 2000, 123, 591-600.	7.6	42
167	Mitochondrial DNA Point Mutation T9176C in Leigh Syndrome. Journal of Child Neurology, 2000, 15, 830-833.	1.4	19
168	A Missense Mutation of Cytochrome Oxidase Subunit II Causes Defective Assembly and Myopathy. American Journal of Human Genetics, 1999, 65, 1030-1039.	6.2	131
169	Cytochrome c Oxidase Deficiency Associated with the First Stop-Codon Point Mutation in Human mtDNA. American Journal of Human Genetics, 1998, 63, 29-36.	6.2	135
170	UK centres are not following the Royal College of Pathologists' recommendations for storage of Guthrie cards: a national policy is needed.. Journal of Medical Genetics, 1998, 35, 263-263.	3.2	6
171	Mitochondrial disorders. Current Paediatrics, 1997, 7, 123-127.	0.2	1
172	Late presentation of biotinidase deficiency with acute visual loss and gait disturbance. Developmental Medicine and Child Neurology, 1997, 39, 830-831.	2.1	25
173	Leigh syndrome: Clinical features and biochemical and DNA abnormalities. Annals of Neurology, 1996, 39, 343-351.	5.3	694
174	Production and disposal of medium-chain fatty acids in children with medium-chain acyl-CoA dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 1994, 17, 74-80.	3.6	19